

Version 2.0



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## *Abstract*

[Back to Hit List](#)**Grant Number:** 1Z01HG000059-01**PI Name:** STRUEWING, JEFFERY**PI Email:****PI Title:****Project Title:** Outcomes of Education and Counseling for HNPCC Testing

**Abstract:** Individuals with colon cancer and their first degree relatives are being offered gene testing for Hereditary Non-Polyposis Colorectal Cancer (HNPCC). Knowledge and expectations are assessed at baseline, followed by pre-test psychological assessment and an education and counseling session. Participants choosing testing receive mutation results in person along with discussion of available surveillance options from 3-6 months after their decision. Psychological and behavioral aspects of their decision making are evaluated and outcomes of the testing process assessed for those not choosing testing as well as those choosing testing at multiple points for up to 1 year post risk notification. The third year of this study has continued to focus on the identification of families with mutations in two of the four known HNPCC genes, MSH2 and MLH1. To date, 221 individuals have been screened for inclusion in the protocol through analysis of mismatch repair in tumors, i.e. RER status. A breakdown of those studies is as follows: RER results pending (30), Receipt of tumor blocks pending (21), RER Positive (33), Off Study (RER negative tumor) (94), Discrepancy between lab interpretation (36). In addition, subject recruitment has been approached through the identification of individuals with colon cancer who have a family history consistent with the Amsterdam criteria for HNPCC. To date, 34 individuals with family medical histories consistent with the Amsterdam criteria have been identified and recruited into the study. Following informed consent, education and counseling, 32 of the 34 have elected to undergo germline testing. Eleven individuals have been identified with mutations (stop codons) in the MSH2/MLH1/MSH6 genes. The identification of these mutations have afforded the opportunity to offer participation of first degree relatives in the study following discussions, approval and consent of the proband. At this point in time, 38 family members have consented to participate. Thirty-seven (37) of the 38 have chosen germline testing. Six and 12 month follow up questionnaires are now being completed. Patient accrual is continuing.

**Thesaurus Terms:**

colon neoplasm, gene mutation, genetic counseling, neoplasm /cancer diagnosis, neoplasm /cancer education, neoplasm /cancer genetics  
cancer risk, family genetics, gene expression, health behavior, prognosis, psychological

adaptation  
human subject

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